

FACT SHEET
Healthcare Provider

Carnitine Uptake Defect, CUD)
Carnitine Transport Defect (CTD)

Description:

Carnitine Uptake Defect is an autosomal recessive disorder in which the body cannot oxidize fatty acids properly. The disorder is caused by a defect in the plasma membrane transport of carnitine. This error results in a deficiency of carnitine, a failure of acylcarnitine formation, and inadequate transport of certain fatty acids into the mitochondria. The mitochondrial β -oxidation pathway plays a major role in energy production, especially during periods of fasting. The ability of the cells to produce energy and to remove toxic wastes is impaired. Carnitine deficiency can be life threatening.

Incidence in General Population:

1:100,000 live births

Symptoms:

Onset of symptoms may occur in two forms: one with onset in infancy, between birth and 30 months of age, and a late onset form between 1 and 7 years of age. Symptoms may include hypoketotic hypoglycemia, cardiomyopathy, muscle weakness, seizures, vomiting, liver dysfunction, and lethargy progressing to coma. If left untreated, death can occur.

Diagnosis:

Newborn Screening—Tandem mass spectrometry identifies reduced concentrations of free carnitine (“C0”). A second dried-blood-spot filter paper card may be requested by the Newborn Screening Laboratory if the initial screening result is above the normal range. Infants with presumptive positive screening (critical) results require prompt follow up. If this occurred, the clinician would be contacted by the Metabolic Treatment Center. When notified of these results, the clinician should immediately check on the clinical status of the baby and facilitate referral to the Metabolic Treatment Center. The Metabolic Treatment Center will provide consultation and assistance with diagnostic testing.

Monitoring:

Plasma levels monitored for free carnitine levels.

Treatment:

Treatment consists of L-carnitine supplementation and avoidance of fasting. L-carnitine can reverse the heart problems and muscle weakness. Infants and young children need to eat often to avoid problems. They should go no more than 4 to 6 hours without food. It is important that they be fed during the night. They need to be awakened if they do not wake up on their own.

Illness and Immunizations:

- Patients with CUD must be monitored closely during times of illness. Conditions that can cause metabolic de-compensation include:
 - Poor appetite
 - Low energy or excessive sleepiness
 - Vomiting
 - Diarrhea
 - Infection
 - Fever
 - Persistent muscle pain or weakness

- Infants and children with CUD need to consume extra-starchy food and drink more fluids during any illness—even if they do not feel hungry. Hospitalization may be necessary.
- Immunizations should be kept current.

Surgical/Surgical Procedures:

Major stresses, such as surgery or accidents, can be complicated for a CUD patient. Extreme care is required to avoid problems during such periods.

- Discuss any plans for surgical and dental procedures with the Metabolic Treatment Center.
- Preoperative fasting should be avoided with 10% dextrose being started preoperatively and continuing postoperatively until the child is eating and drinking well.
- If complicated surgery or a postoperative period as an inpatient is required, the procedure should be done at a hospital with a metabolic service.

Growth and Development:

It is crucial to closely monitor all growth parameter on a regular basis.



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